Soft Bones
Finding the Key to HPP

How to talk to your family about your HPP diagnosis

Sharing results of your HPP diagnosis with family members is a personal decision. That being said, you are a source of important health information for your close blood relatives. The information can help other family members with their own healthcare planning.

If you've recently been diagnosed with hypophosphatasia (HPP), you may have undergone genetic testing to identify the mutation associated with HPP. As a genetic disease, HPP is passed along in families. If one family member is diagnosed, many times additional family members also have the HPP genetic mutation, which may or may not cause symptoms.

Talking to family members about the possibility of an HPP diagnosis should be done with care, especially since the impact of this information can have different reactions from different people. For example, an expecting family member may find this information troubling and be concerned about the health of their unborn child. Other family members, who are asymptomatic, may be in denial or, in some cases, feel a sense of guilt for their role in passing along a genetic disease.

Although challenging to discuss, communication is key and important for many reasons. A family member may already be exhibiting symptoms but hasn't connected it to an HPP diagnosis. Another family member may be considering having children and would benefit from seeking genetic counseling to help inform next steps and prepare for what to expect. Still others may be genetic carriers of the mutation, and while they may not have any symptoms currently, they could develop symptoms later in life.

Inherited Diseases and Genetic Testing

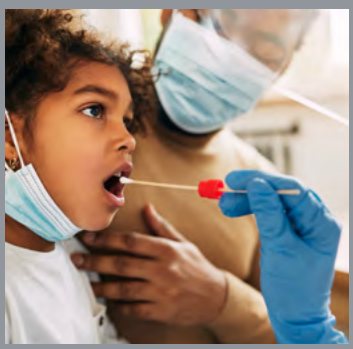
Genetic testing provides information about your genes and their potential impact on overall health. Genetic testing identifies abnormalities that may be present that increase the risk of developing certain diseases. The results may have implications not only for you but also for blood relatives about their likelihood of developing a genetic disorder, like HPP. It may feel overwhelming and unclear as to how to talk to family members about your genetic testing results. It's important to have these discussions, even though they are challenging as your results can impact your family members. There is no right formula or way to have these discussions that works best for everyone. It's up to you, and the particular dynamics of your family, to determine how much information you share, when, and with whom.

The purpose of this discussion guide is to help you prepare for a productive dialogue with family members who may be at risk.

Hypophosphatasia (HPP) is a genetic condition caused by one or more mutations (changes) in the ALPL gene resulting in low levels of the enzyme alkaline phosphatase. ALP is necessary for the healthy development of teeth and bones. When ALP is low, bones and teeth are unable to absorb the minerals and calcium that are critical for strength and hardness. As a result, bones are weak, soft and prone to fractures. Teeth often fall out prematurely.

Low ALP levels are the hallmark of HPP. There are reliable laboratory tests that measure ALP levels in the bloodstream and, supported by other diagnostic tests and a clinical exam by a healthcare provider, can be used to confirm an HPP diagnosis.

Genetic testing can support a diagnosis and help you better understand your family inheritance pattern of HPP. Because HPP can be inherited as an autosomal recessive trait (among brothers and sisters) or an autosomal dominant trait (multiple generations), genetic testing is often recommended to determine if other family members are at increased risk and can help you both plan for the future and guide treatment decisions.



Considerations for Genetic Testing

If you haven't had genetic testing, and are considering it for HPP, there are pros and cons to consider.

The good news is, it's easier than ever to undergo genetic testing. Although there still may be some challenges with insurance coverage, many reliable commercial labs offer this testing directly to consumers and there are other options available. Some commercial labs that screen for HPP do require a healthcare provider to authorize and order a genetic test, while others may allow individuals to request a test online. Some labs may be covered by insurance, while others may not. Test results are always strictly confidential. If you are uncertain about where to go for testing or which test is best for you, ask your physician or reach out to us at info@softbones.org.

Considerations for Sharing Genetic Test Results with Family Members

Should I share my HPP diagnosis with my family?

Many people recognize that knowing their family health history is important, especially their risk for developing certain heritable health conditions. Sharing this information can help family members guide their own care and planning, now or in the future. In addition, there may be signs and symptoms of HPP that family members will want to be aware of, so they can monitor them closely.

Who should I tell?

Since HPP is a genetic disease, members of your direct family (those who share the same bloodline, like siblings) would be a good place to start.

What is the best way to share the information?

The goal is to share information without creating unnecessary tension. Be prepared that not all family members will want to discuss medical history or genetic risk for HPP. Some may not be ready to receive this information all at once. Be respectful and understand that some people may need time to process the information you are providing. Be open to the conversation when it arises.

It is very helpful to share your personal journey with your family members. Having a clear understanding of your personal process, in which you articulate the symptoms that lead to your diagnosis can be incredibly powerful. It is important to note: rarely do two HPP patients experience the exact same symptoms, so another family member's experience may be completely different. Highlight this as part of the discussion.

Sharing a resource with a basic disease overview is also a great way to educate family members about the disease. This provides facts without an emotional component. Sometimes, this is an easier way for people to process new, and challenging, information.

Preparing for your discussion

Receiving life-changing information is challenging for anyone. Although you want to try to stick to the facts when sharing, you need to be prepared for the potential emotional reactions you may receive.

Most importantly, validate the feelings being expressed. It is understandable that you may feel defensive or frustrated by another's reaction. It is important to validate what they are saying and how they are reacting. Below are some common emotions that might be expressed and some validating ways to respond.



Scenarios for Practice and Discussion

Please note, this discussion guide is intended to help prepare for potential scenarios and not a script. Feel free to adapt talking points to your own style.

ANGER

When people feel threatened or overwhelmed, they often respond with misdirected anger. Learning that they may have a genetic predisposition to a particular illness can cause this type or response. Rather than defend yourself, take some deep breaths, validate the anger and concern and encourage more conversation at a later date.

I understand that this is a lot of information to take in and can see that you are angry and frustrated about what I am telling you. Rather than fight about it now, let's take some time and come back to the conversation later.

FEAR/WORRY

When we feel uncertain, our fear and worry are triggered. Our default when things feel out of our control is to feel anxious. Learning that you may be a carrier of HPP can trigger worries about how it will affect one's life. When you are met with worry, do your best to continue to focus on the facts.

I hear that you are anxious and that this is triggering a lot of worrisome thoughts. Since we don't have all the information yet, let's try to focus on what we do know now.

GUILT

Guilt is an emotion that is often unjustified. We experience it when we feel responsible for an outcome. A genetic predisposition is no one's fault.

I can see that you feel that this is your fault or responsibility. Genetics influence so much of who we are and what we are, and they started a long time before you! Let's talk about what we can do moving forward.

DENIAL/DISBELIEF

Similarly to anger and anxiety, when we are told challenging information, our immediate reaction may be to deny its existence. This is a self-protective mechanism that helps in the short-term and not in the long-term. Validation is so important to slow down the reactivity. You may not be able to convince anyone of anything in an emotional moment, and will, most likely, have to come back to it at a later time.

I know that this sounds like it cannot be true and that I made it all up. It's a lot to take in, especially when it's not something you have heard about before. I have information and facts that I can provide you around this when you are ready to look it over with me. I'm also happy to meet with you and the doctor so you can learn more and have a better understanding of what this all means.

SADNESS

Scary and overwhelming news can trigger sadness for many. Learning that you may have HPP or be a genetic carrier can cause an increase in sadness as it does have an impact on many areas of one's life. Rather than try to convince someone not to be sad, join them in the feeling supporting them as they work through it.

It is a lot of information to take in and it may feel like you don't have the options in your life that you thought you did. We don't really know all the answers yet and your sadness about it makes so much sense. I'm here to support you in whatever way I can.

Remember: you are not responsible for another person's reactions. Nor are you responsible for changing their reactions. You are only responsible for your own reactions. The better YOU respond, the better the interaction will be.

What if family members have questions I can't answer?

You aren't expected to be a medical or genetics expert. Your role is simply to provide them with information about HPP as it pertains to you and connect them with appropriate resources so that they can learn their own histories and become their own advocates.

Genetics and inheritance patterns are very complicated. Within families, it is important to discuss them as needed, so members can manage their own medical needs as best as possible. While these discussions can be challenging, they are vital in helping people access the help they need when they need it.

Below are some helpful HPP resources.

Resources

Information on genetics and genetic testing can be found on the U.S. Centers for Disease Control and Prevention (CDC) website here.

For more information on genetic counseling or to find a certified genetic counselor, visit the National Society of Genetic Counselors at www.nsgc.org.

To learn more about the genetics of HPP, please visit the Soft Bones website here.

For more information on the Genetic Information Nondiscrimination Act, visit the Department of Labor's GINA website here.

Information about GINA and other laws can also be found at the National Human Genome Research Institute website here.

Reach out to info@softbones.org if you need additional information or support.

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